Michigan Department of Community Health
MDCH
Jennifer M. Granholm, Governor
Janet Olszewski, Director

www.mi.gov/newbornscreening

Michigan Resources & Support

Children's Hospital of Michigan Metabolic Clinic
Toll-free: 1-866-44CHMMC

Children's Special Health Care Services
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www.earlychildhoodmichigan.org

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E-mail: MDCH-newbornscreening @ michigan.gov

Michigan NBS Parent Liaison
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E-mail: NBS-parent@michigan.gov

National Resources & Support

Biotinidase Family Support Group
www.biotinidasedeficiency.20m.com/

Connecticut Children's Medical Cntr.
www.ccmckids.org/research/biotinidase

Family Village
www.familyvillage.wisc.edu

GeneReviews
www.genetests.org

Genetic Alliance
www.geneticalliance.org

Revised Dec. 2005

What is Biotinidase Deficiency?

Biotinidase deficiency is an inherited disorder preventing babies from using biotin (a needed vitamin) in a normal manner. Biotin is required to break down certain foods. Biotinidase deficiency occurs in about 1 in 27,000 Michigan newborns. If untreated, profound biotinidase deficiency can result in seizures, mental retardation or coma. The milder form called partial biotinidase deficiency is not associated with serious complications.

How may Biotinidase Deficiency affect my child?

Timing of Signs and Symptoms

Usually signs of biotinidase deficiency occur in infancy or early childhood. Signs may occur from the first few weeks of life until 10 years of age or older.

Common Early Signs

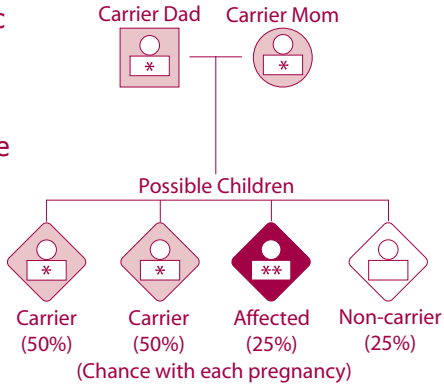
- Seizures
- Low muscle tone (hypotonia)
- Hair loss (alopecia)
- Skin rashes

Common Later Signs

- Developmental delay
- Speech problems
- Vision and hearing loss
- Abnormalities in movement and muscle control (Ataxia)

How does Biotinidase Deficiency occur?

Biotinidase deficiency is a genetic disorder. Parents of an affected child carry a genetic trait that can cause biotinidase deficiency. Both parents pass the trait to a child with biotinidase deficiency. There is a 1 in 4 chance that each child will have biotinidase deficiency when both parents carry the trait for the disorder.



How is Biotinidase Deficiency treated?

Newborns and children are given biotin (vitamin) supplements. No special diet is required. Regular monitoring and care through the Metabolic Clinic and your pediatrician are required to ensure proper health, growth and development.

For more information contact the Newborn Screening Program toll-free at 1-866-852-1247 or e-mail NBS-Parent@michigan.gov

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University of Michigan Health System- Pediatric Endocrinology
Phone: 734-764-5175

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National Resources & Support

CAH Parent Handbook
www.dshs.state.tx.us/newborn/cah.shtm

CAH Printable Booklet
www.med.jhu.edu/pedendo/cah

CARES Foundation, Inc.
Toll-free: 1-866-227-3737
www.caresfoundation.org

Family Village
www.familyvillage.wisc.edu

GeneReviews
www.genetests.org

Genetic Alliance
www.geneticalliance.org

What is Congenital Adrenal Hyperplasia (CAH)?

CAH is an inherited disorder present at birth that affects a baby's adrenal glands. A baby with CAH cannot make the proper amount of certain steroid hormones. CAH occurs in about 1 in 18,000 Michigan babies. Without treatment, problems with growth and development will occur and some babies may also become ill and die.

How may CAH affect my child?

Symptoms in Girls

The abnormal adrenal gland may affect an unborn baby girl. Some baby girls are born with masculinized (boy-like in appearance) external genitalia. Girls with CAH usually have normal internal reproductive organs. Girls with untreated CAH may not go through normal puberty.

Symptoms in Boys

Boys with CAH appear normal at birth. Boys with untreated CAH may go through puberty at a very early age.

Salt-Wasting CAH

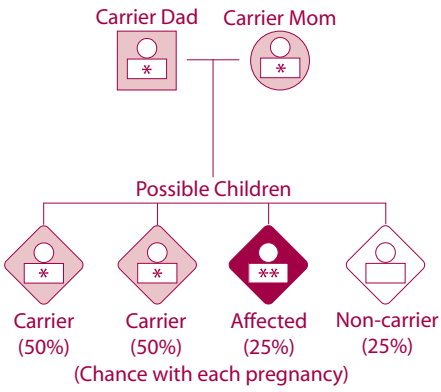
A severe form of CAH is called "Salt-Wasting" CAH. A baby with this form of CAH loses large amounts of salt (sodium) in the urine. This can lead to symptoms such as:

- Dehydration
- Muscle weakness
- Poor growth
- Vomiting
- Weakness of the heart

Babies with untreated salt-wasting CAH may become ill and die in infancy.

How does CAH occur?

CAH is a genetic disorder. Parents of an affected child carry a genetic trait causing CAH. Both parents pass the trait to a child with CAH. There is a 1 in 4 chance that each child will have CAH when both parents carry the trait for the disorder.



How is CAH treated?

Newborns are placed on steroid hormones as soon as possible. These hormones must be taken for a person's entire lifetime. Children with the salt-wasting form of CAH may also require salt supplements. Reconstructive plastic surgery on external genitalia may be needed for some newborn baby girls with CAH. Regular visits to a doctor specializing in diseases that affect hormones (endocrinologist) are needed to monitor the amount of medicine required by a child. Your pediatric endocrinologist and pediatrician will ensure your child is prescribed the proper amount of medicine for his or her own unique needs and growth.

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Family Village
www.familyvillage.wisc.edu

Galactosemic Families of Minnesota
www.galactosemia-mn.com

GeneReviews
www.genetests.org

Genetic Alliance
www.geneticalliance.org

Parents of Galactosemic Children, Inc.
www.galactosemia.org

Revised Dec. 2005

What is Galactosemia?

Galactosemia is an inherited disorder that prevents a baby from digesting a sugar (galactose) found in milk and most infant formulas (except soy). Classic galactosemia occurs in about 1 in 41,000 Michigan newborns. If untreated, galactose can build up in a baby's body causing damage to the eyes, liver and brain. Galactosemia can be life threatening in the first few weeks of life. Milder variants of galactosemia (Duarte) are not associated with serious complications.

How may Galactosemia affect my child?

Classic Galactosemia

Signs of classic galactosemia are usually present in the first two weeks of life. These signs may include:

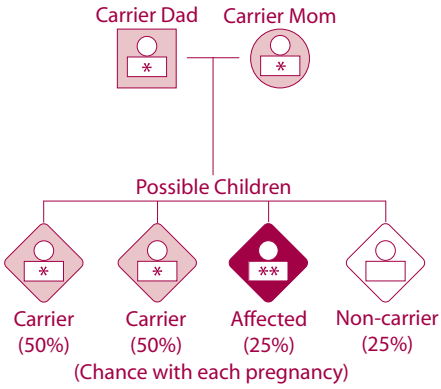
- Vomiting and diarrhea
- Poor weight gain or poor suck
- Jaundice (yellowing of skin or whites of eyes)
- Enlarged liver
- Cataracts
- Nervous system damage

Duarte Galactosemia Variant

Children with this form of galactosemia usually do not have any signs or symptoms. Follow-up in the Metabolic Clinic is recommended to monitor growth and development.

How does Galactosemia occur?

Galactosemia is a genetic disorder. Parents of an affected child carry a genetic trait that can cause galactosemia. Both parents pass the trait to a child with galactosemia. There is a 1 in 4 chance that each child will have galactosemia when both parents carry the trait for the disorder.



How is Galactosemia treated?

Newborns are placed on a special formula restricting galactose. This formula consists of a milk (lactose) substitute that is usually soy. Blood monitoring of galactose and strict avoidance of dairy products will allow for the best possible outcome in children with galactosemia. The Metabolic Clinic and your pediatrician will help you begin and keep your child on a diet that meets his or her metabolic needs.

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GeneReviews
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Genetic Alliance
www.geneticalliance.org

National Coalition for PKU and Allied Disorders
Toll-free: 1-877-996-2723
www.pku-allieddisorders.org

STAR-G Project
www.newbornscreening.info/Parents/facts.html

Revised Dec. 2005

What is Homocystinuria?

Homocystinuria is an inherited disorder in which the baby is unable to digest part of a protein found in food and milk. It occurs in about 1 in 200,000 newborns. Without treatment, babies with homocystinuria will have problems with bone development, learning, vision and blood clotting.

How may Homocystinuria affect my child?

Developmental Delay and Mental Retardation

Developmental delay is often the first sign of homocystinuria in the untreated child. Newborn screening and early treatment shortly after birth will offer a child the best outcome.

Bone (skeletal) Problems

An untreated child may be taller than family members. Bones may be longer than usual. About one-half of untreated children will also develop weak bones (osteoporosis) by their teens.

Eye (ocular) Problems

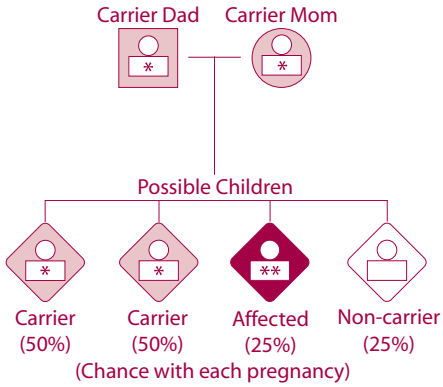
A dislocated lens in the eye (ectopia lentis) is usually seen in untreated children by 8 years of age. Near-sightedness (myopia) is also common.

Blood Clotting Problems

Untreated children may develop a blood clot (thromboembolism) in any blood vessel. In untreated young adults, these blood clots may lead to an early death.

How does Homocystinuria occur?

Homocystinuria is a genetic disorder. Parents of an affected child carry a genetic trait causing homocystinuria. Both parents pass the trait to a child with homocystinuria. There is a 1 in 4 chance that each child will have homocystinuria when both parents carry the trait for the disorder.



How is Homocystinuria treated?

Newborns are placed on a special formula to restrict protein. Children must have frequent metabolic monitoring. Special formulas and vitamin supplements including vitamin B6 may be given. The Metabolic Clinic and your pediatrician will help you begin and keep your child on a diet that meets his or her metabolic needs.

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Family Village
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FOD Family Support Group
Phone: 336-547-8682
www.fodsupport.org

GeneReviews
www.genetests.org

Genetic Alliance
www.geneticalliance.org

STAR-G Project
www.newbornscreening.info/Parents/facts.html

Revised Dec. 2005

What is MCAD Deficiency?

(Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency)

MCAD deficiency is an inherited fatty acid oxidation disorder that interferes with the body's ability to use fat as an energy source. This may occur after vigorous exercise, missing a meal or when fighting a simple infection like the stomach flu. MCAD deficiency occurs in about 1 in 26,000 Michigan newborns. Without treatment, babies with MCAD deficiency may suffer seizures, coma or even sudden death.

How may MCAD Deficiency affect my child?

Most babies born with MCAD deficiency are healthy. Their bodies cannot process certain types of fat to make energy when the normal sugar energy source, glucose, runs out.

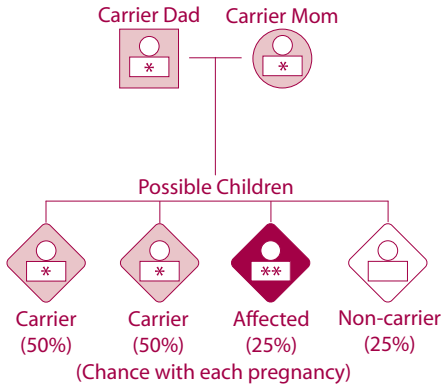
Most children will have signs of the disorder between 2 months and 2 years of age. Some babies may have signs of MCAD deficiency as early as 2 days after birth. It is sometimes found in adults who have only mild signs of MCAD deficiency and did not know they were affected.

A child with MCAD deficiency may develop the following health problems during a time of illness, lack of eating or vigorous exercise:

- Vomiting
- Lethargy (extreme tiredness)
- Seizures
- Breathing difficulties
- Heart failure
- Coma

How does MCAD Deficiency occur?

MCAD deficiency is a genetic disorder. Parents of an affected child carry a genetic trait causing MCAD deficiency. Both parents pass the trait to a child with MCAD deficiency. There is a 1 in 4 chance that each child will have MCAD deficiency when both parents carry the trait for the disorder.



How is MCAD Deficiency treated?

MCAD deficiency is treated by avoiding periods of not eating. Most metabolic specialists recommend that babies under 3 months of age not go without food for more than 4 hours. A visit to the emergency room for intravenous sugar may be needed during times of illness or if the child is unable to eat. A high fat diet should be avoided. Some doctors may treat children with a medication called L-Carnitine. The Metabolic Clinic and your pediatrician will work with you and your child to ensure a proper diet is begun and continued to allow for normal health, growth and development.

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STAR-G Project
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Revised Dec. 2005

What is Maple Syrup Urine Disease (MSUD)?

MSUD is an inherited disorder in which the baby is unable to use parts of protein found in food and milk. It occurs in about 1 in 235,000 Michigan newborns. Without treatment, babies with MSUD may become seriously ill and die before they are diagnosed.

How may MSUD affect my child?

Symptoms in the Untreated Newborn

Babies with MSUD often begin to show signs of the disorder within the first few days of life. Symptoms in the untreated newborn include:

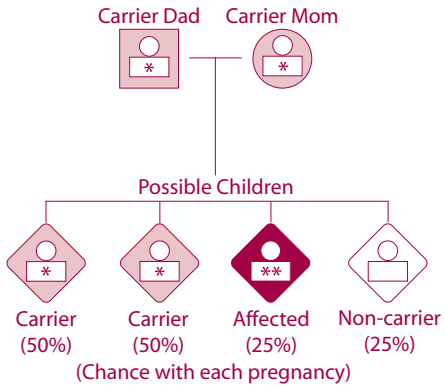
- Poor suck or poor feeding
- Vomiting
- Maple syrup odor
- Lethargy (tiredness)
- Rigid muscle tone
- Seizures
- Brain injury
- Eventual coma and death

Developmental Delay and Mental Retardation

If a child with MSUD is not treated, life threatening metabolic crisis can occur. This may cause damage to the brain, resulting in mental retardation. Children diagnosed early in life before severe symptoms appear have a chance for normal intelligence with strict diet care.

How does MSUD occur?

MSUD is a genetic disorder. Parents of an affected child carry a genetic trait causing MSUD. Both parents pass the trait to a child with MSUD. There is a 1 in 4 chance that each child will have MSUD when both parents carry the trait for the disorder.



How is MSUD treated?

Newborns are placed on a special formula to restrict protein. A diet restricting protein should be continued through life and must be carefully monitored. A child with vomiting, diarrhea or decreased liquid intake must receive medical care immediately. Regular monitoring and care through the Metabolic Clinic and your pediatrician are required to ensure your child's health, growth and development.

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Family Village
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Genetic Alliance
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National Urea Cycle Disorders Foundation
Toll-Free: 1-800-386-8233
www.nucdf.org

STAR-G Project
www.newbornscreening.info/Parents/facts.html

Revised Dec. 2005

What are Citrullinemia and Argininosuccinic Acidemia (ASA)?

Citrullinemia and ASA are inherited, urea cycle disorders that result when a baby's body cannot remove certain waste products from the blood. These disorders occur in about 1 in 250,000 newborns. Without treatment, progressive brain damage and death occur.

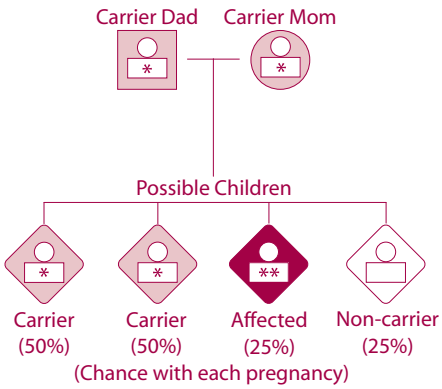
How may Citrullinemia and ASA affect my child?

Babies with citrullinemia and ASA cannot process ammonia, a by-product of the proteins we eat, into a harmless product called urea. This causes ammonia to build up in their blood. High levels of ammonia are toxic (poisonous) and cause the symptoms of citrullinemia and ASA. These symptoms in babies include:

- Loss of appetite
- Vomiting
- Listlessness (Loss of energy/movement)
- Seizures

How do Citrullinemia and ASA occur?

Citrullinemia and ASA are genetic disorders. Parents of an affected child carry a genetic trait causing citrullinemia or ASA. Both parents pass the trait to an affected child. There is a 1 in 4 chance that each child will have citrullinemia when both parents carry the trait for the disease. There is a 1 in 4 chance that each child will have ASA when both parents carry the trait for the disease.



How are Citrullinemia and ASA treated?

Newborns are placed on a special formula to restrict protein. Certain supplements and medicines may be given. Kidney dialysis may be needed for some children. The Metabolic Clinic and your pediatrician will help you begin and keep your child on a diet that meets his or her metabolic needs.

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What is Phenylketonuria (PKU)?

PKU is an inherited disorder in which the baby is unable to use a certain part of protein (phenylalanine) found in food and milk. PKU occurs in about 1 in 8,800 Michigan newborns. Without treatment, phenylalanine will build up in a baby's body and mental retardation will result.

How may PKU affect my child?

Developmental Delay and Mental Retardation

Mental retardation will occur if a child with PKU does not begin a special diet within the first 7 to 10 days of life. If the special diet is started at the correct time and followed properly, a child should not have mental retardation due to PKU.

Diet

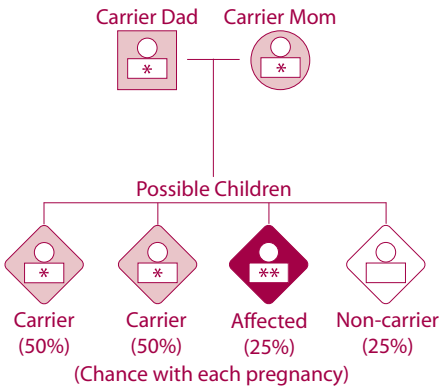
A special formula restricting phenylalanine must be started as soon as possible. Special food and formula are available for a child with PKU. The special diet should be followed throughout life.

Pregnancy

It is vital for a woman with PKU to understand the importance of staying on her special diet before becoming pregnant. A woman who becomes pregnant while off of her diet has a high risk for having a baby with mental retardation and birth defects. Continued use of the special diet will allow a normal chance for healthy children.

How does PKU occur?

PKU is a genetic disorder. Parents of an affected child carry a genetic trait causing PKU. Both parents pass the trait to a child with PKU. There is a 1 in 4 chance that each child will have PKU when both parents carry the trait for the disorder.



How is PKU treated?

Newborns are placed on a special formula to restrict protein. The Metabolic Clinic will help you begin and keep your child on a special diet that meets his or her metabolic needs. Frequent monitoring of your child's phenylalanine level is needed to make sure it is in the safe range. The Metabolic Clinic and your pediatrician will help you with all aspects of your child's PKU care. A support parent is also available to help with practical tips on raising a child with PKU.

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www.geneticalliance.org

March of Dimes
www.marchofdimes.com

National Coalition for PKU and Allied Disorders
Toll-free: 1-877-996-2723
www.pku-allieddisorders.org

Revised Dec. 2005

Michigan Department of Community Health
MDCH
Jennifer M. Granholm, Governor
Janet Olszewski, Director

www.mi.gov/newbornscreening

Michigan Resources & Support

Children's Special Health Care Services
Family Phone Line
Toll-free: 1-800-359-3722

Early On® Michigan
Toll-free: 1-800-EARLY ON
www.earlychildhoodmichigan.org

Michigan Genetics Connection
www.migeneticsconnection.org

Michigan Newborn Screening
Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: MDCH-newbornscreening @ michigan.gov

Michigan NBS Parent Liaison
Toll-free: 1-866-852-1247
E-mail: NBS-parent@michigan.gov

University of Michigan Health System - Pediatric Endocrinology
Phone: 734-764-5175

National Resources & Support

Family Village
www.familyvillage.wisc.edu

GeneReviews
www.genetests.org

The MAGIC Foundation
www.magicfoundation.org/www/docs/114.5

What is Congenital Hypothyroidism?

Congenital hypothyroidism is a thyroid disorder present at birth. The disorder occurs when a baby's thyroid gland does not make enough of the hormone, thyroxine. This hormone is needed for normal growth and good health. Congenital hypothyroidism occurs in about 1 in 1,900 Michigan newborns. Without treatment mental retardation and poor growth may result.

How may Congenital Hypothyroidism affect my child?

Signs of congenital hypothyroidism in a newborn may be very subtle. Some babies do not develop signs until they are a few months old. Signs of congenital hypothyroidism in a newborn include:

- Feeding problems
- Lethargy (Lack of energy)
- Jaundice (Yellowing of skin or whites of eyes)
- Constipation
- Larger protruding tongue
- Cold mottled skin
- Sluggish reflexes

How does Congenital Hypothyroidism occur?

Congenital hypothyroidism is usually sporadic, meaning it occurs by chance in a child. Sometimes a specific cause for a child's congenital hypothyroidism can be found such as:

- Absent or small thyroid gland
- A thyroid hormone abnormality
- A rare genetic (inherited) form
- Maternal medication use (rare)

The following medical professionals may be able to help find a cause for your child's congenital hypothyroidism:

- Endocrinologist
- Clinical Geneticist
- Pediatrician

How is Congenital Hypothyroidism treated?

Immediate diagnosis and treatment for a baby with congenital hypothyroidism are needed in the newborn period. Treatment with thyroid hormone is usually effective if started in the first few weeks of a baby's life. The hormone is given in a tablet form that will need to be crushed in formula or breast milk. The tablet should not be mixed with soy formula or formula with iron. Regular monitoring and care by your endocrinologist and pediatrician are required to ensure your baby's good health, growth, and development.

For more information contact the Newborn Screening Program toll-free at 1-866-852-1247 or e-mail NBS-Parent@michigan.gov

Supported in part by project #5 H91MC00215-03-00 as a Special Project of Regional and National Significance (SPRANS), Title V (as amended), Social Security Act, administered by the Maternal and Child Health Bureau, Health Resources and Services Administration, United States Department of Health and Human Services.

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Michigan Resources & Support

Children's Hospital of Michigan Sickle Cell Clinic
Phone: 313-745-5613

Children's Special Health Care Services
Family Phone Line
Toll-free: 1-800-359-3722

Early On® Michigan
Toll-free: 1-800-EARLY ON
www.earlychildhoodmichigan.org

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E-mail: MDCH-newbornscreening @ michigan.gov

Michigan NBS Parent Liaison
Toll-free: 1-866-852-1247
E-mail: NBS-parent@michigan.gov

Sickle Cell Disease Association, Michigan Chapter
Toll-free: 1-800-842-0973
www.sicklecelldisease.org

National Resources & Support

About Sickle Cell Disease
www.sicklecellinfo.net

American Sickle Cell Anemia Association
Phone: 216-229-8600
www.ascaa.org/

Family Village
www.familyvillage.wisc.edu

GeneReviews
www.genetests.org

Genetic Alliance
www.geneticalliance.org

Revised Dec. 2005

What is Sickle Cell Anemia?

Sickle cell anemia is an inherited disorder that affects a baby's red blood cells. Babies with sickle cell anemia have fewer red blood cells (anemia) and abnormally shaped red blood cells. Sickle cell anemia can occur in any ethnic group but is more common in babies of African American heritage. It occurs in about 1 in 600 African American babies. Early detection and treatment will help many babies with sickle cell anemia avoid sudden death from infections.

How may Sickle Cell Anemia affect my child?

Abnormal Red Blood Cells

Anemia: Babies and children with sickle cell anemia have chronic (constant) anemia. Signs of anemia include tiredness, paleness and shortness of breath.

Abnormally Shaped Red Blood Cells: Babies with sickle cell anemia have red blood cells shaped like a "sickle", or crescent instead of being round. These cells may damage the spleen. This prevents the body from fighting infection as well as it should.

Pain

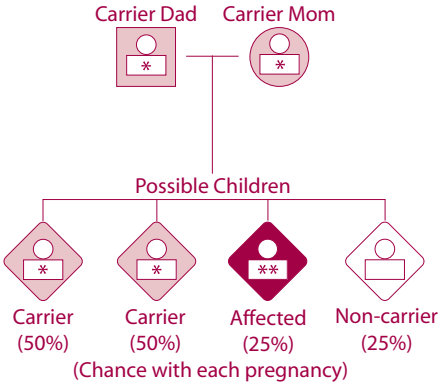
Painful swelling of hands and feet is one of the first signs of sickle cell anemia. Episodes of joint or organ pain may continue throughout a person's lifetime.

Delayed Growth/Puberty

Children with sickle cell anemia often have delays in growth and puberty due to their anemia.

How does Sickle Cell Anemia occur?

Sickle Cell Anemia is a genetic disorder. Parents of an affected child carry a genetic trait that can cause sickle cell anemia. Both parents pass the trait to a child with sickle cell anemia. There is a 1 in 4 chance in each pregnancy that a child will have sickle cell anemia when both parents carry the trait for the disorder.



How is Sickle Cell Anemia treated?

It is essential that children up to 5 years of age receive daily doses of penicillin to help prevent infection that could cause early death. Fever, which could mean a life threatening infection, should be treated as a medical emergency. The child's parent or guardian should know what to do when fever occurs and have a written emergency care plan. Regular care by a sickle cell clinic and/or pediatrician is recommended.

For more information contact the Newborn Screening Program toll-free at 1-866-852-1247 or e-mail NBS-Parent@michigan.gov

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